

Clinical Cytogenetics

Aug. 31th to Sep. 7th, 2019

Goldrain, South Tyrol, Italy

DIRECTOR:

A. Schinzel (Zurich, Switzerland)

FACULTY:

D. Bartholdi (Bern, Switzerland), A. Baumer (Zürich, Switzerland), P. Benn (Farmington CT, U.S.A.), R. Ciccone (Pavia, Italy), Jean-Michel Dupont (Paris, France), E. Klopocki (Würzburg, Germany), T. Krones (Zurich, Switzerland), K. Madan (Leiden, the Netherlands), K. Miller (Hannover, Germany), Rolph Pfundt (Nijmegen, The Netherlands), E. Syk-Lundberg (Stockholm, Sweden), G. van Buggenhout (Leuven, Belgium), O. Zuffardi (Pavia, Italy)

PROVISIONAL PROGRAM:

Saturday, August 31 th		Arrival at Goldrain
19:00-20:30	<i>Dinner</i>	
20:30-21:00	A. Schinzel	Comments on purpose and content of the course
21:00-	Get together	Steinkeller
Sunday, September 1 st		Basic and clinical cytogenetics
Morning Session: 09:00 – 12:40		
09:00-09:35	A. Schinzel	Introduction and organizational comments
09:40-10:15	1 K. Miller	Basic concepts of chromosome aberrations and epidemiology
10:20-10:55	2 D. Bartholdi	Genetic counseling for chromosome aberrations
11:00-11:20	<i>Coffee Break</i>	
11:25-12:00	3 T. Krones	Ethical issues
12:05-12:40	4 D. Bartholdi	Clinical dysmorphic examination in cytogenetic disorders
12:45-14:00	<i>Lunch Break</i>	
Afternoon Session: 14:00 – 18:30		Clinical cytogenetics
14:00-14:35	5 A. Schinzel	Dysmorphic findings in chromosome aberrations: formation and interpretation
14:40-15:15	6 E. Syk-Lundberg	Small supernumerary chromosomes
15:20-15:55	7 K. Miller	Ring chromosomes
16:00-16:20	<i>Coffee Break</i>	
16:25-17:10	W1 D. Bartholdi	Clinical issues: difficult situations
	W2 T. Krones	Ethical issues: difficult situations

17:15-17:45	8	K. Miller	Cytogenetics of spontaneous abortions and stillbirths
17:50-18:35	W1	D. Bartholdi	Clinical issues: difficult situations
	W2	T. Krones	Ethical issues: difficult situations
19:00-20:00		<i>Dinner</i>	
21:00-		Get together	Steinkeller

Monday, September 2nd	Molecular cytogenetic techniques
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Morning Session: 09:00 – 12:40

09:00-09:35	9	K. Miller, E. Klopocki	ISCN, standard and molecular
09:40-10:15	10	A. Baumer	MLPA and QF-PCR
10:20-10:55	11	J.M. Dupont	FISH techniques and their application
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	12	R. Pfundt	SNP array analysis and interpretation in constitutional genome diagnostics
12:05-12:40	13	R. Ciccone	Array: principles, technical aspects, different platforms
12:45-14:00		<i>Lunch Break</i>	

Afternoon Session: 14:00 – 18:40	Continuation and practical exercises
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14:00-14:35	14	K. Madan	Cytogenetic polymorphisms
14:40-15:10	15	O. Zuffardi	Balanced and unbalanced genomic rearrangements: mechanisms of origin
15:15-15:50	16	A. Baumer	Uniparental disomy (UPD) and imprinting
15:55-16:30	17	K. Madan	Inversions and insertions
16:35-16:50		<i>Coffee Break</i>	
16:50-17:25	W3	K. Miller	Practical exercises in report writing. Parallel with W4
	W4	R. Ciccone/ E. Klopocki	Databases for the interpretation of array results for beginners: hands-on in computer room. Parallel with W3
17:30-18:05	18	O. Zuffardi	Genomic mosaicism and diseases
18:10-18:45	W3	K. Miller	Practical exercises in report writing. Parallel with W4
	W4	R. Ciccone/ E. Klopocki	Databases for the interpretation of array results for beginners: hands-on in computer room. Parallel with W3
19:00-20:00		<i>Dinner</i>	
21:00-		Get together	Steinkeller

Tuesday, September 3rd	Prenatal diagnosis
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Morning Session: 09:00 – 12:40

09:00-09:35	19	P. Benn	Traditional approaches for the detection of aneuploidy in prenatal diagnosis: screening using serum and ultrasound markers
09:40-10:15	20	J.M. Dupont	Prenatal cytogenetic diagnosis I: mosaics in PND
10:20-10:55	21	J.M. Dupont	Prenatal cytogenetic diagnosis II: RAD (rapid aneuploidy detection) and CMA (chromosomal microarray detection)
11:00-11:20		<i>Coffee Break</i>	

11:25-12:00	22	P. Benn	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects I
12:05-12:40	23	P. Benn	Non-invasive prenatal aneuploidy testing using cfDNA: current status and future prospects II

12:45-14:00 *Lunch Break*

Afternoon Session: 14:10 – 18:30 EXCURSION

19:00-20:00	<i>Dinner</i>	
21:15-	Get together	Steinkeller

Wednesday, September 4th Prenatal Diagnosis, miscellaneous

Morning Session: 09:00 – 12:40

09:00-09:45	24	E. Syk-Lundberg	Pre-implantation genetic diagnosis
09:50-10:25	25	J. Wisser	Prenatal ultrasound findings indicative for chromosome aberrations
10:30-11:05	26	O. Zuffardi	Incomplete penetrance and genomic rearrangements
11:10-11:30	<i>Coffee Break</i>		
11:30-12:05	27	G.v. Buggenhout	Clinical dysmorphic examination in cytogenetic disorders. II
12:10-12:45	28	A. Schinzel	Twins at prenatal diagnosis
12:45-14:00	<i>Lunch Break</i>		

Afternoon Session: 14:00 – 18:30 Practical exercises; miscellaneous

14:00-14:35	29	P. Benn	Origin of trisomy
14:40-15:15	30	E. Klopocki	Position effects and effects of CNVs containing no genes
15:20-15:55	31	O. Zuffardi	The legacy of trisomy rescue: from imprinting diseases to de novo unbalanced chromosome rearrangements
15:55-16:15	<i>Coffee Break</i>		
16:15-17:00	W5	A. Baumer	Work-up of cases: strategies after the detection of CNVs. Parallel with W6
	W6	R. Ciccone/ E. Klopocki	Databases for the interpretation of array results for advances users: hands-on in computer room. Parallel with W5
17:05-17:40	32	K. Madan	Mosaics and chimeras
17:45-18:30	W6	R. Ciccone/ E. Klopocki	Databases for the interpretation of array results for advances users: hands-on in computer room. Parallel with W5
	W5	A. Baumer	Work-up of cases: strategies after the detection of CNVs. Parallel with W6
19:00-20:30	<i>Dinner</i>		
21:00-	Get together	Steinkeller	

Thursday, September 5th	Clinical cytogenetics
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Morning Session: 09:00 – 12:40	
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09:00-09:35	33	A. Schinzel	Clinical phenotypes in UPD
09:40-10:15	34	G. v. Buggenhout	Microdeletion syndromes
10:20-10:55	35	K. Madan	Complex chromosome rearrangements
11:00-11:20		<i>Coffee Break</i>	
11:25-12:00	36	G. v. Buggenhout	Common chromosome disorders and follow-up I
12:05-12:40	37	G. v. Buggenhout	Common chromosome disorders and follow-up II
12:45-14:10		<i>Lunch Break</i>	

Afternoon Session: 14:15 – 18:40	Sex chromosome aberration; miscellaneous
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14:10-14:45	38	G.v.Buggenhout	The clinical effect of sex chromosomal aneuploidy
14:50-15:25	39	O. Zuffardi	Sex chromosomes: mosaics, AZF deletions, XIST, SRY
15:30-16:05	40	K. Madan	Disorders of sex development
16:05-16:25		<i>Coffee Break</i>	
16:25-17:10	W7	K. Miller	Segregation of chromosomal translocations: practical exercises
17:10-17:55	W7	K. Miller	Segregation of chromosomal translocations: practical exercises (half of the group, rotate with W7)
18:00-18:40	41	O. Zuffardi	Why are certain rearrangements more frequent than others?
19:00-20:00		<i>Dinner</i>	
20:15-		Get together	Steinkeller

Friday, September 6th	Miscellaneous
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Morning Session: 09:00 – 12:40	New techniques
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09:00-09:50	42	R. Pfundt	Next generations sequencing strategies in diagnostics to detect nucleotide and structural variants in the human genome
09:55-10:50	43	E. Klopocki	An introduction to CRISPR/Cas9 and its future possibilities
10:55-11:15		<i>Coffee Break</i>	
11:15-12:40	44	Students	Student presentations
12:45-14:10		<i>Lunch Break</i>	

Afternoon Session: 14:15 – 18:45	Second excursion or self-study
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19:00-20:00		<i>Dinner</i>	
20:15-		Get together	Steinkeller

Saturday, September 7st	Past and future of clinical cytogenetics
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Morning Session: 09:00 – 12:40	
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09:00-09:40	W9	P.Benn	Appropriate management in prenatal screening and diagnosis
09:45-10:20	45	O. Zuffardi	From cytogenetics to cytogenomics
10:25-11:00	46	XX	Future of human cytogenetics, prenatal

11:05-11:20		<i>Coffee Break</i>	
11:20-11:55	47	O. Zuffardi	Future of human cytogenetics, postnatal
12:00-12:35	48	A. Schinzel	Paradigm shifts, unexpected findings and use of markers in cytogenetics

12:40-14:00 *Lunch Break*

Afternoon Session: 14:00 – 18:45	Evaluation; examination; castle tour
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14:00-16:30	49	Students / Faculty	Student presentations and discussion of the program
16:30-16:50		<i>Coffee Break</i>	
16:50-18:00		Students	EXAMINATION
18:00-18:45		Students / Faculty	Guided visit of the castle
19:30-21:00		<i>Farewell dinner</i>	Steinkeller or Inner Court of the castle

Sunday, September 2nd	Goodbye Goldrain
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08:00-09:00	<i>Breakfast</i>
09:00-	<i>Checkout and departure</i>